JAK3 gene

Janus kinase 3

Normal Function

The *JAK3* gene provides instructions for making a protein that is critical for the normal development and function of the immune system. The JAK3 protein is part of a signaling pathway called the JAK/STAT pathway, which transmits chemical signals from outside the cell to the cell's nucleus. Signals relayed by the JAK3 protein regulate the growth and maturation of certain types of white blood cells (lymphocytes) called T cells and natural killer cells. In addition, JAK3 is important for the normal maturation of another type of lymphocyte called B cells. T cells, B cells, and natural killer cells attack bacteria, viruses, and fungi and help regulate the entire immune system.

Health Conditions Related to Genetic Changes

JAK3-deficient severe combined immunodeficiency

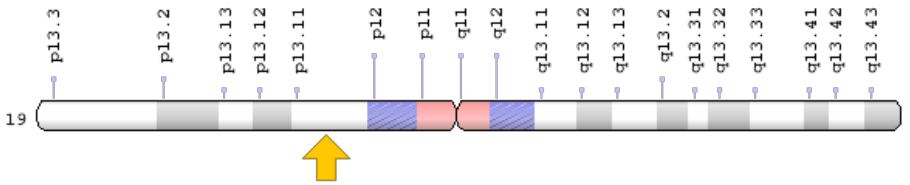
More than 50 *JAK3* gene mutations have been identified in people with *JAK3*-deficient severe combined immunodeficiency (SCID), an impairment of the immune system that leads to repeated and persistent infections that can be life-threatening.

Most mutations in the *JAK3* gene prevent the production of JAK3 protein or result in a protein that is unstable and quickly broken down. Others lead to production of a protein that cannot perform its function. Without functional JAK3 protein, certain chemical signals are not relayed to the nucleus, impairing lymphocyte growth and development. As a result, individuals with *JAK3*-deficient SCID have few or no T cells and natural killer cells and a normal number of poorly functioning B cells. The shortage of functional lymphocytes causes people with this condition to be susceptible to infections.

Chromosomal Location

Cytogenetic Location: 19p13.11, which is the short (p) arm of chromosome 19 at position 13.11

Molecular Location: base pairs 17,824,782 to 17,848,071 on chromosome 19 (Homo sapiens Updated Annotation Release 109.20200522, GRCh38.p13) (NCBI)



Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- JAK-3
- JAK3_HUMAN
- JAKL
- Janus kinase 3 (a protein tyrosine kinase, leukocyte)
- L-JAK
- leukocyte Janus kinase
- LJAK
- tyrosine-protein kinase JAK3

Additional Information & Resources

Educational Resources

- Immunobiology: The Immune System in Health and Disease (fifth edition, 2001): Defects in T-cell Function Result in Severe Combined Immunodeficiencies https://www.ncbi.nlm.nih.gov/books/NBK27109/#A1509

Scientific Articles on PubMed

- PubMed https://www.ncbi.nlm.nih.gov/pubmed?term=%28%28JAK3%5BTIAB%5D%29+OR+%28Janus+kinase+3%5BTIAB%5D%29%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+720+days%22%5Bdp%5D

Catalog of Genes and Diseases from OMIM

- JANUS KINASE 3
  http://omim.org/entry/600173

Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology
  http://atlasgeneticsoncology.org/Genes/JAK3ID41032ch19p13.html
- ClinVar
  https://www.ncbi.nlm.nih.gov/clinvar?term=JAK3%5Bgene%5D
- HGNC Gene Symbol Report
  https://www.genenames.org/data/gene-symbol-report/#!/hgnc_id/HGNC:6193
- Monarch Initiative
  https://monarchinitiative.org/gene/NCBIGene:3718
- NCBI Gene
  https://www.ncbi.nlm.nih.gov/gene/3718
- UniProt
  https://www.uniprot.org/uniprot/P52333

Sources for This Summary

- OMIM: JANUS KINASE 3
  http://omim.org/entry/600173
- Notarangelo LD, Mella P, Jones A, de Saint Basile G, Savoldi G, Cranston T, Vihinen M, Schumacher RF. Mutations in severe combined immune deficiency (SCID) due to JAK3 deficiency. Hum Mutat. 2001 Oct;18(4):255-63. Review.
  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/11668610
- O'Shea JJ, Husa M, Li D, Hofmann SR, Watford W, Roberts JL, Buckley RH, Changelian P, Candotti F. Jak3 and the pathogenesis of severe combined immunodeficiency. Mol Immunol. 2004 Jul;41(6-7):727-37. Review.
  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/15220007
- Pesu M, Candotti F, Husa M, Hofmann SR, Notarangelo LD, O'Shea JJ. Jak3, severe combined immunodeficiency, and a new class of immunosuppressive drugs. Immunol Rev. 2005 Feb;203: 127-42. Review.
  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/15661026
- Vihinen M, Villa A, Mella P, Schumacher RF, Savoldi G, O'Shea JJ, Candotti F, Notarangelo LD. Molecular modeling of the Jak3 kinase domains and structural basis for severe combined immunodeficiency. Clin Immunol. 2000 Aug;96(2):108-18.
  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/10900158

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